(19) World Intellectual Property Organization International Bureau





(43) International Publication Date 18 December 2003 (18.12.2003)

PCT

(10) International Publication Number WO 03/104398 A2

(51) International Patent Classification7:

C12N

- (21) International Application Number: PCT/US03/17262
- (22) International Filing Date: 4 June 2003 (04.06.2003)
- (25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

60/385,901

6 June 2002 (06.06.2002) US

- (71) Applicant (for all designated States except US): JOHNS HOPKINS UNIVERSITY SCHOOL OF MEDICINE [US/US]; Suite 906, 111 Market Place, Baltimore, MD 21202 (US).
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): YAN, Hai [CN/US]; 7236 Montgomery Road, apartment #3c, Elkridge, MD 21075 (US). KINZLER, Kenneth, W. [US/US]; 1403 Halkirk Way, Bel Air, MD 21015 (US). VOGELSTEIN, Bert [US/US]; 3700 Breton Way, Baltimore, MD 21208 (US).

- (74) Agent: KAGAN, Sarah, A.; Banner & Witcoff, Ltd., 1001 G Street, N.W., Eleventh Floor, Washington, DC 20001 (US).
- (81) Designated States (national): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NI, NO, NZ, OM, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.
- (84) Designated States (regional): ARIPO patent (GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO, SE, SI, SK, TR), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

 without international search report and to be republished upon receipt of that report

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

03/104398 A2

(54) Title: ALLELIC VARIATION IN HUMAN GENE EXPRESSION

(57) <u>Abstract:</u> Genetically-determined variation in expression levels is an important component of human diversity and has significant implications for normal and abnormal human physiology. Using this genetically determined variation one can identify disease risk factors in individuals. One can associate such variations with birth defects, diseases, and non-disease traits. Such variations can be associated with susceptibility or resistance to the effects of drugs or other therapeutic interventions.